

AI-based pathology predicts origins for cancers of unknown primary

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Abstract

Cancer of unknown primary (CUP) origin is an enigmatic group of diagnoses in which the primary anatomical site of tumour origin cannot be determined^{1,2}. This poses a considerable challenge, as modern therapeutics are predominantly specific to the primary tumour³. Recent research has focused on using genomics and transcriptomics to identify the origin of a tumour^{4–9}. However, genomic testing is not always performed and lacks clinical penetration in low-resource settings. Here, to overcome these challenges, we present a deep-learning-based algorithm—Tumour Origin Assessment via Deep Learning (TOAD)—that can provide a differential diagnosis for the origin of the primary tumour using routinely acquired histology slides. We used whole-slide images of tumours with known primary origins to train a model that simultaneously identifies the tumour as primary or metastatic and predicts its site of origin. On our held-out test set of tumours with known primary origins, the model achieved a top-1 accuracy of 0.83 and a top-3 accuracy of 0.96, whereas on our external test set it achieved top-1 and top-3 accuracies of 0.80 and 0.93, respectively. We further curated a dataset of 317 cases of CUP for which a differential diagnosis was assigned. Our model predictions resulted in concordance for 61% of cases and a top-3 agreement of 82%. TOAD can be used as an assistive tool to assign a differential diagnosis to complicated cases of metastatic tumours and CUPs and could be used in conjunction with or in lieu of ancillary tests and extensive diagnostic work-ups to reduce the occurrence of CUP. A deep-learning-based algorithm uses routinely acquired histology slides to provide a differential diagnosis for the origin of the primary tumour for complicated cases of metastatic tumours and cancers of unknown primary origin.